Priv.-Doz. Dr. rer. nat. Barbara Vona

GENERAL INFORMATION

Address: Institute for Auditory Neuroscience
University Medical Center Göttingen
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Current position: Group leader of the Hearing Genomics Group at the Institute for Auditory Neuroscience & InnerEarLab

ACADEMIC EDUCATION

2011-2014	PhD Human Genetics, University of Würzburg
2006-2008	Master of Science, Pathobiology, University of Arizona (Tucson, AZ, USA)
2002-2006	Bachelor of Science, Microbiology, University of Arizona (Tucson, AZ, USA)

SCIENTIFIC DEGREES

2022	Umhabilitation, Human Genetics, University Medical Center Göttingen
2019	Habilitation, Human Genetics, University of Würzburg (Prof. T. Haaf)
2014	PhD (Dr. rer. nat.), Human Genetics, University of Würzburg (Prof. T. Haaf)
2008	Master of Science in Pathobiology, University of Arizona (Tucson, AZ, USA)
2006	Bachelor of Science in Microbiology, University of Arizona (Tucson, AZ, USA)

PROFESSIONAL CAREER AFTER COMPLETING DEGREE

Since 2025	Visiting scientist, Harvard Medical School, Boston, MA, USA
Since 2025	Visiting scientist, The Broad Institute, MIT & Harvard, Cambridge, MA, USA
Since 2024	Accepted into the Heisenberg Program (German Research Foundation)
Since 2022	Group Leader
2021-2022	Fellow, Institute for Auditory Neuroscience & InnerEarLab, University Medical Center Göttingen
2018-2021	Junior Group Leader, Tübingen Hearing Research Center, University of Tübingen
2014-2018	Postdoctoral fellow, Institute of Human Genetics, University of Würzburg

MISCELLANEOUS Further Scientific Activities *(selected)*

Since 2026	International Rare Diseases Research Consortium (IRDiRC) Task Force on Optimizing the Use of Data Sources and Registries
Since 2026	Nominated by the German Society of Human Genetics as an expert for amending the pediatrics directive for newborn hearing screening in Germany
Since 2025	Nominated by the German Society of Human Genetics to amend the German S2k Guidelines for Cochlear Implant Treatment
Since 2025	Coordinator of the ClinGen Splicing Working Group
Since 2025	Member of the ClinGen/AVE (Atlas of Variant Effects) Functional Data Working Group
Since 2025	Scientific Advisory Board for the Cure KARS – Laia Foundation
Since 2025	Associate Editor, Molecular Syndromology
Since 2024	Establishment of the Center for Rare Hearing Disorders within the Centers for Rare Diseases framework at UMG
Since 2024	Medical and Scientific Advisory Board of The SPATA Foundation
Since 2023	Developer and study manager for the “Hereditary Hearing Impairment Patient Registry” for patients with Otoferlin (*OTOF*) and *CABP2* associated hearing impairment. ClinicalTrials.gov IDs: NCT05946057, NCT06680934

Since 2023	Member of the Atlas of Variant Effects Alliance
Since 2022	Associate Editor, Journal of the Association for Research in Otolaryngology
Since 2022	Genomics England PanelApp Gene Reviewer for Auditory Neuropathy
Since 2022	Genomics England PanelApp Gene Reviewer for Hearing Loss
Since 2021	Member of the Consortium for Gene Diagnostics
Since 2019	Member of the ClinGen Hearing Loss Variant Curation Expert Panel
Since 2019	Member of the ClinGen Hearing Loss Gene Curation Expert Panel

SELECTED PUBLICATIONS *(with scientific assurance)*

1. Ni C*, Wei Y*, **Vona B***, Park D, Wei Y, Schmitz DA, Ding Y, Sakurai M, Ballard E, Li L, Liu Y, Kumar A, Xing C, Qin S, Kim S, Foglizzo M, Zhao J, Kim HG, Ekmekci C, Karimiani EG, Imannezhad S, Eghbal F, Badv RS, Schwaibold EMC, Dehghani M, Vahidi Mehrjardi MY, Metanat Z, Eslamiyeh H, Khouj E, Nasser Alhajj SM, Chedrawi A, Ramzan K, Hashmi JA, Alluqmani MM, Basit S, Veltra D, Marinakis NM, Niotakis G, Vorgia P, Sofocleous C, Lee H, Jeong WC, Umair M, Bilal M, Pinheiro Ferreira Alves CA, Sieber M, Kruer M, Houlden H, Alkuraya FS, Zeqiraj E, Greenberg RA, Cenik C, Yu L, Maroofian R, Wu J, Buszczak M. (2025) A programmed decline in ribosome levels governs human early neurodevelopment. Nat Cell Biol 27(8):1240-1255
2. **Vona B**[#] (2025) Rethinking non-syndromic hearing loss and its mimics in the genomic era. Eur J Hum Genet Eur J Hum Genet 33(2):147-150.
3. **Vona B**, Wollnik B, Strenzke N, Moser T (2024) Catching up but still miles behind-a patient registry for otoferlin. Exp Mol Med 56(6):1472-1473.
4. Redfield SE*, De-la-Torre P*, Zamani M, Wang H, Khan H, Morris T, Shariati G, Karimi M, Kenna MA, Seo GH, Xu H, Lu W, Naz S, Galehdari H, Indzhykulian AA**[,#], Shearer AE**[,#], **Vona B**[**,#] (2024) PKHD1L1, a gene involved in the stereocilia coat, causes autosomal recessive nonsyndromic hearing loss. Hum Genet 143(3):311-329.
5. Lin SJ*, **Vona B***, Lau T, Huang K, Zaki MS, Aldeen HS, Karimiani EG, Rocca C, Noureldeen MM, Saad AK, Petree C, Bartolomaeus T, Abou Jamra R, Zifarelli G, Gotkhindikar A, Wentzensen IM, Liao M, Cork EE, Varshney P, Hashemi N, Mohammadi MH, Rad A, Neira J, Toosi MB, Knopp C, Kurth I, Challman TD, Smith R, Abdalla A, Haaf T, Suri M, Joshi M, Chung WK, Moreno-De-Luca A, Houlden H, Maroofian R**, Varshney GK** (2023) Evaluating the association of biallelic OGDHL variants with significant phenotypic heterogeneity. Genome Med 15(1):102.
6. **Vona B***, Schwartzbaum DA**, Rodriguez AA**, Lewis SS, Toosi MB, Radhakrishnan P, Bozan N, Akın R, Doosti M, Manju R, Duman D, Sineni CJ, Nampoothiri S, Karimiani EG, Houlden H, Bademci G, Tekin M*, Girisha KM*, Maroofian R*, Douzgou S* (2022) Biallelic KITLG variants lead to a distinct spectrum of hypomelanosis and sensorineural hearing loss. J Eur Acad Dermatol Venereol 36(9):1606-1611.
7. Lin SJ*, **Vona B***, Barbalho PG*, Kaiyrzhanov R, Maroofian R, Petree C, Severino M, Stanley V, Varshney P, Bahena P, Alzahrani F, Alhashem A, Pagnamenta AT, Aubertin G, Estrada-Veras JI, Hernández HAD, Mazaheri N, Oza A, Thies J, Renaud DL, Dugad S, McEvoy J, Sultan T, Pais LS, Tabarki B, Villalobos-Ramirez D, Rad A; Genomics England Research Consortium, Galehdari H, Ashrafzadeh F, Sahebzamani A, Saeidi K, Torti E, Elloumi HZ, Mora S, Palculict TB, Yang H, Wren JD, Ben Fowler, Joshi M, Behra M, Burgess SM, Nath SK, Hanna MG, Kenna M, Merritt JL 2nd, Houlden H, Karimiani EG, Zaki MS, Haaf T, Alkuraya FS, Gleeson JG, Varshney GK (2021) Biallelic variants in KARS1 are associated with neurodevelopmental disorders and hearing loss recapitulated by the knockout zebrafish. Genet Med 23(10):1933-1943.
8. **Vona B**[#], Mazaheri N, Lin SJ, Dunbar LA, Maroofian R, Azaiez H, Booth KT, Vitry S, Rad A, Rüschendorf F, Varshney P, Fowler B, Beetz C, Alagramam KN, Murphy D, Shariati G, Sedaghat A, Houlden H, Petree C, VijayKumar S, Smith RJH, Haaf T, El-Amraoui A*, Bowl MR*[,#], Varshney GK*, Galehdari H* (2021) A biallelic variant in CLRN2 causes nonsyndromic hearing loss in humans. Hum Genet 140(6):915-931.

9. Lin YC*, Niceta M*, Muto V*, **Vona B***, Pagnamenta AT, Maroofian R, Beetz C, van Duyvenvoorde H, Dentici ML, Lauffer P, Vallian S, Ciolfi A, Pizzi S, Bauer P, Grüning NM, Bellacchio E, Del Fattore A, Petrini S, Shaheen R, Tiosano D, Halloun R, Pode-Shakked B, Albayrak HM, Işık E, Wit JM, Dittrich M, Freire BL, Bertola DR, Jorge AAL, Barel O, Sabir AH, Al Tenaiji AMJ, Taji SM, Al-Sannaa N, Al-Abdulwahed H, Digilio MC, Irving M, Anikster Y, Bhavani GSL, Girisha KM; Genomics England Research Consortium, Haaf T, Taylor JC, Dallapiccola B, Alkuraya FS, Yang RB, Tartaglia M (2021) SCUBE3 loss-offunction causes a recognizable recessive developmental disorder due to defective bone morphogenetic protein signaling. Am J Hum Genet 108(1):115-133.
10. Rad A, Schade-Mann T, Gamerdinger P, Yanus GA, Schulte B, Müller M, Imyanitov EN, Biskup S, Löwenheim H, Tropitzsch A, **Vona B**[#] (2021) Aberrant COL11A1 splicing causes prelingual autosomal dominant nonsyndromic hearing loss in the DFNA37 locus. Hum Mutat 42(1):25-30.

* equal contribution, ** equal contribution, [#] corresponding author